

TABLE 1. Sequences of amplicon-specific primers.

| Cystic Fibrosis Transmembrane Regulator (CFTR) 15-plex               |              |                  | Gauchers (GCR) and Sickle Cell Anemia (SCA) 4-plex  |            |                  |
|--|--------------|------------------|---|------------|------------------|
| Primer Sequences   | Exon         | Size (bp)        | GCR Primer Sequences  | Exon       | Size (bp)        |
| AGG CTT CTC AGT GAT CTG TTG<br>GAA TCA TTC AGT GGG TAT AAG CAG       | Int 19       | ~440             | GGG TGG GAG GGT GGA GGC TAA TGG<br>CCA GAA GGT AGA AAG GTG AG   | 6          | 401              |
| GCC CGA CAA ATA ACC AAG TGA<br>AGT CTA ACA AAG CAA GCA GTG           | 19           | 410              | GAA TGT CCC AAG CCT TTG A<br>AAG CTG AAG CAA GAG AAT CG   | 2          | 358              |
| TGA TGG TAA GTA CAT GGG TG<br>CAA AAG TAC CTG TTG CTC CA             | 21           | 381              | TGC AAC TAC TGA GGC ACT T<br>TAC AAT GAT GGG ACT GTC G  | 9          | 319              |
| CTT CTA ATG GTG ATG ACA GCC T<br>CCA CTG AAA ATA ATA TGA GGA AAT     | 9            | 335              | SCA Primer Sequences  |            |                  |
| AGG TAG CAG CTA TTT TTA TGG<br>TAA GGG AGT CTT TTG CAC AA            | 13           | 295              | CAT TTG CTT CTG ACA CAA CTG<br>CCA ACT TCA TCC ACG TTC ACC  |            | 124              |
| TGT AGG AAG TCA CCA AAG<br>CGA TAC AGA ATA TAT GTG CC                | 4            | 267              | GCR and Tay-Sachs (TS) 3-plex   |            |                  |
| GGA GTC CAA TTT TCA CTC ATC TTG<br>AGT TAA TGA GTT CAT AGT ACC TGT T | 17b          | 245              | GCR<br>CCT TGC CCT GAA CCC CGA A<br>CTG ACT CTG TCC CTT TAA TGC CCA   | 9, 10, 11  | 871              |
| AGA TAC TTC AAT AGC TCA GCC<br>GGT ACA TTA CCT GTA TTT TGT TT        | 7            | 220              | TS Primer Sequences<br>GTG TGG CGA GAG GAT ATT CCA<br>TGG CTA GAT GGG ATT GGG TCT   | 11, 12***  | 530              |
| CAG ATT GAG CAT ACT AAA AGT G<br>TAC ATG AAT GAC ATT TAC AGC A       | 11           | 200              | GGG TCC TAC AAC CCT GTC ACC CAC<br>AAG CTT CAC TCT GAG CAT AAC AAG  | 7**        | 190              |
| GAG CCT TCA GAG GGT AAA AT<br>TCA CAT AGT TTC TTA CCT CT             | 10           | 175              | B-thalassemia Primer Sequences  |            |                  |
| AAG AAC TGG ATC AGG GAA GA<br>TCC TTT TGC TCA CCT GTG GT             | 20           | 155              | GCT GTC ATC ACT TAG ACC TC<br>GCA AGA AAG CGA GCT TAG TG  | 1, 2, 3    | 1612             |
| GCT GTC AAG CCG TGT TCT A<br>GTA TAA TTT ATA ACA ATA GTG CC          | 5            | 132              | WT-1 Primer Sequences   |            |                  |
| TTG GTT GTG CTG TGG CTC CT<br>ACA ATA CAT ACA AAC ATA GTG G          | 14b          | 110              | CTG AGT GAA TGG AGC GGC<br>GGG TGA ATG AGT AGG TGG  | Name<br>B* | Size (bp)<br>204 |
| GAC TCT CCT TTT GGA TAC CTA<br>GCA TGA GCA TTA TAA GTA AGG           | 12           | 90               | CGG TGC TGG ACT TTG CG<br>AAG TGG ACA GTG AAG GCG   | F          | 186              |
| GGC GAT GTT TTT TCT GGA GA<br>ACA AAT GAG ATC CTT ACC CC             | 3            | 70               | CCG TCT TGC GAG AGC ACC<br>CTA ATT TGC TGT GGG TTA GG   | H*         | 262              |
| CFTR Exon 21 Primer Sequences  |              |                  | AGT TGT GTA TAT TTG TGG TTA TG<br>GTT ACT GTG GAA AGG CAA TG  | J          | 167              |
| CAA GTG AAT CCT GAG CGT GAT TT<br>CAA AAG TAC CTG TTG CTC CA         | Name<br>SS#1 | Size (bp)<br>477 | GAG ATC CCC TTT TCC AG<br>CAC AGC TGC CAG CAA TG  | N*         | 176              |
| GAA CTT GAT GGT AAG TAC ATG GGT G<br>AGT CAA AAG TAC CTG TTG CTC CAG | SS#2         | 389              | CTC ACT GTG CCC ACA TTG<br>CAA TTT CAT TCC ACA ATA G  | O*         | 211              |
| TGA TGG TAA GTA CAT GGG TG<br>CAA AAG TAC CTG TTG CTC CA             | SS#3         | 381              | * Reported previously by Varanasi et al 1994.<br>**Reported previously by Navon & Proia 1989.<br>***Reported previously by Tanaka et al 1990. |            |                  |
|  |              |                  | NOTE:<br>Amplicon sizes increase by 40bp for chimeric primers.  |            |                  |

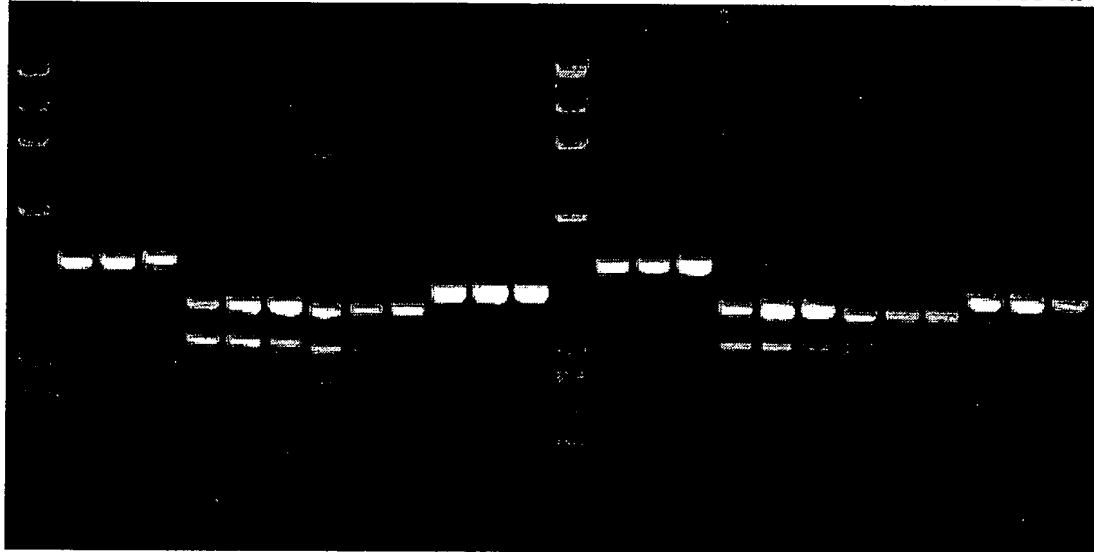
FIGURE 1

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50°

55°

| 50°  |   |   |   |      |   |   |   |      |   |    |    | 55°      |   |   |   |      |   |   |   |      |   |   |    |      |    |  |  |          |  |  |  |
|------|---|---|---|------|---|---|---|------|---|----|----|----------|---|---|---|------|---|---|---|------|---|---|----|------|----|--|--|----------|--|--|--|
| SS#1 |   |   |   | SS#2 |   |   |   | SS#3 |   |    |    | SS#3+UPS |   |   |   | SS#1 |   |   |   | SS#2 |   |   |    | SS#3 |    |  |  | SS#3+UPS |  |  |  |
| M    | 1 | 2 | 3 | 4    | 5 | 6 | 7 | 8    | 9 | 10 | 11 | 12       | M | 1 | 2 | 3    | 4 | 5 | 6 | 7    | 8 | 9 | 10 | 11   | 12 |  |  |          |  |  |  |



60°

65°

| 60°  |   |   |   |      |   |   |   |      |   |    |    | 65°      |   |   |   |      |   |   |   |      |   |   |    |      |    |  |  |          |  |  |  |
|------|---|---|---|------|---|---|---|------|---|----|----|----------|---|---|---|------|---|---|---|------|---|---|----|------|----|--|--|----------|--|--|--|
| SS#1 |   |   |   | SS#2 |   |   |   | SS#3 |   |    |    | SS#3+UPS |   |   |   | SS#1 |   |   |   | SS#2 |   |   |    | SS#3 |    |  |  | SS#3+UPS |  |  |  |
| M    | 1 | 2 | 3 | 4    | 5 | 6 | 7 | 8    | 9 | 10 | 11 | 12       | M | 1 | 2 | 3    | 4 | 5 | 6 | 7    | 8 | 9 | 10 | 11   | 12 |  |  |          |  |  |  |

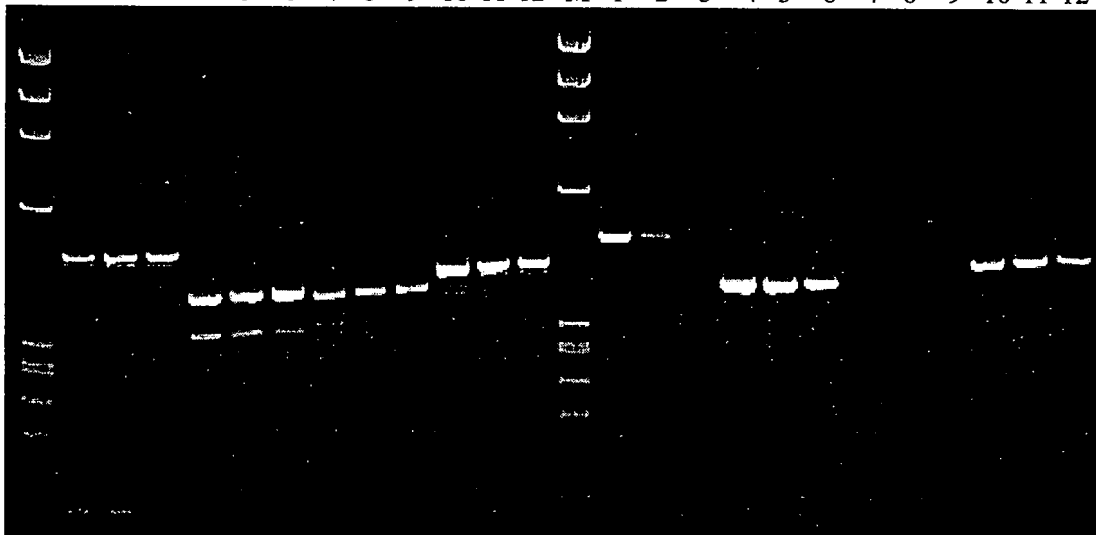


FIGURE 2

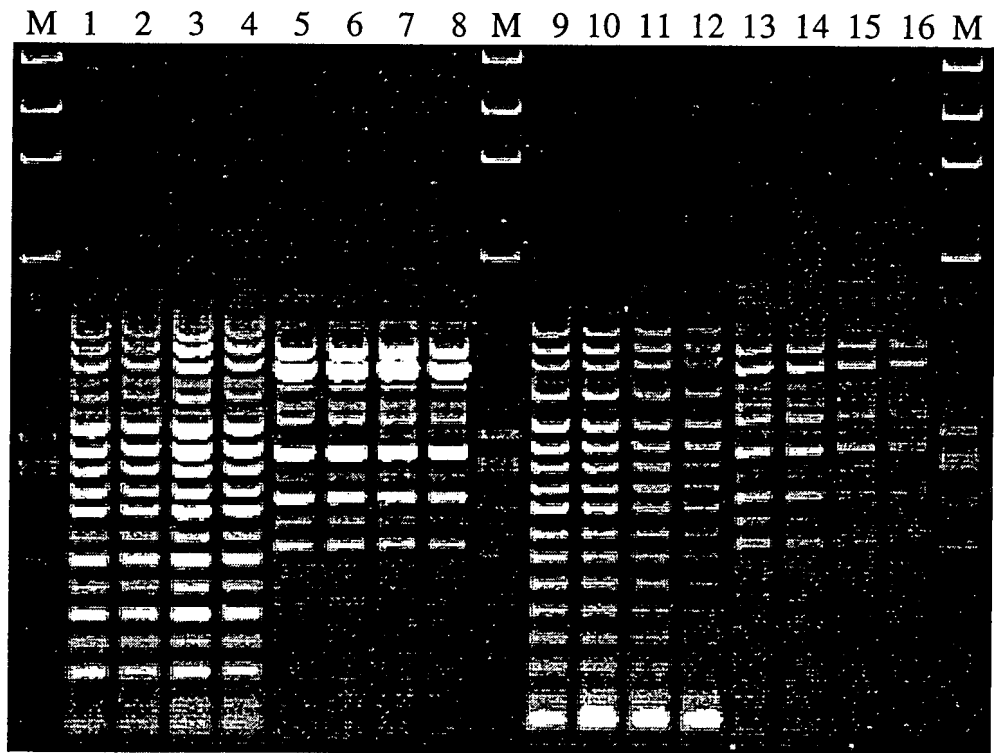


FIGURE 3

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1 2 3 4 5 6 7 8 9 10 11 12 13 14 15 16 17 18 19 20 21 22 23 24 M

fourty

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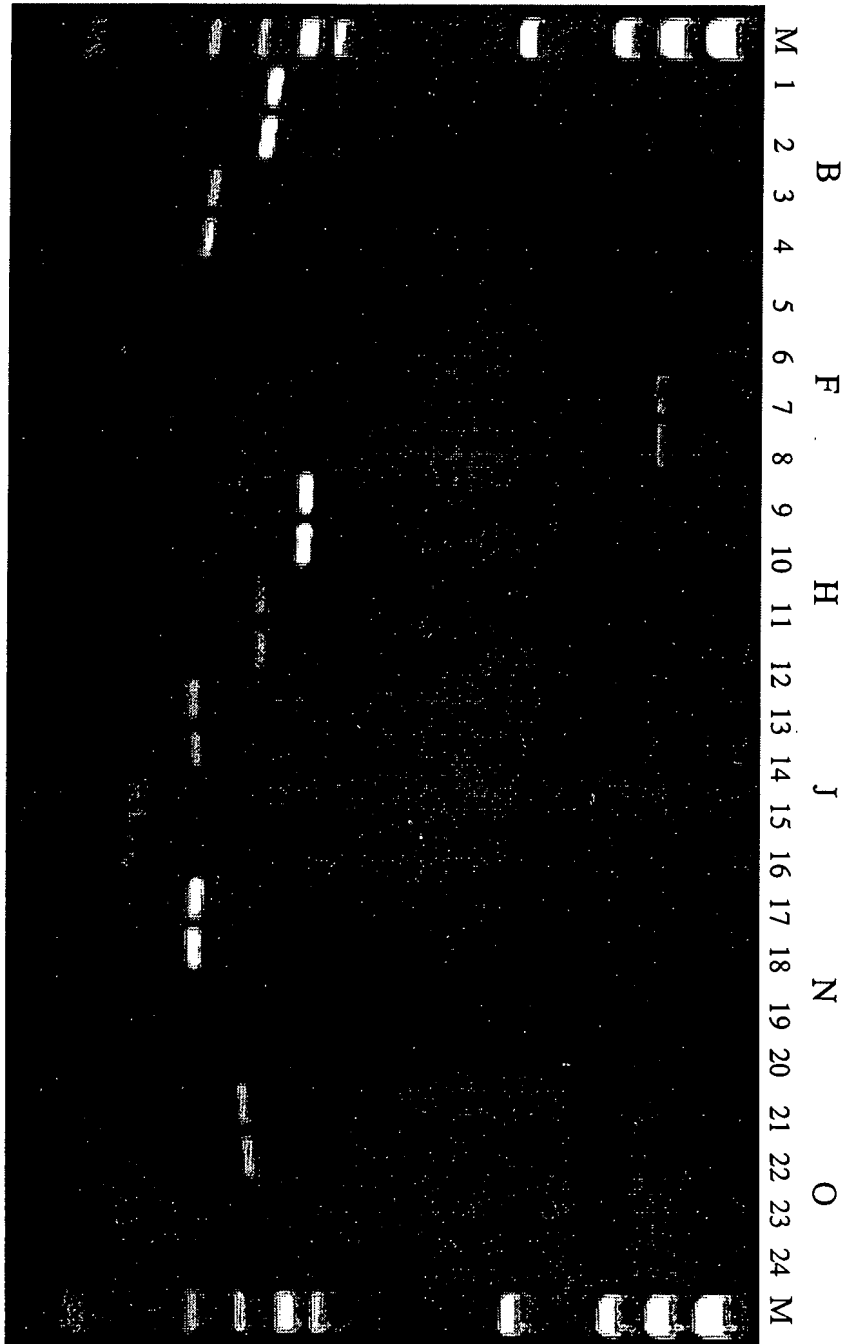


FIGURE 5